Reviewer’s report

Title: Case reports: three Novel variants in PCCA and PCCB Genes in Chinese Patients with propionic acidemia

Version: 1 Date: 07 Jan 2020

Reviewer: Nani Maharani

Reviewer's report:

General comments:

This manuscript is a case report of three Chinese patients with propionic acidemia, of which harbouring three novel mutations in either PCCA or PCCB gene. I have some questions to answer, and suggestions to improve this manuscript:

Abstract section:

- Line 34 (new sentence) - 38 is better to be divided into 2 sentences: 1st sentence describing the mutations in PCCA gene, 2nd sentence in PCCB gene.

- Line 38 (new sentence), suggested revision: ”Among those mutations, three mutations (…please mention) are novel.”

Case presentation:

- How were the patients' condition after the specific diets had been given? Did they show improvement on clinical parameters?

- Methods:

- The reference sequence for PCCA and PCCB gene should be mentioned

- Line 115: Direct Sanger sequencing? For which parts were the sequence performed (coding exons and exon-intron boundaries?) Did the authors already mentioned all the primers being used (including the primers to detect the mutation in the introns)? The primer-design software must also be mentioned.

- Line 168 - 171: please explain the sentence, in which it is mentioned that the frameshift alteration after codon 120 leading to PTC which is located at codon 160.
The compound heterozygous mutation in Patient 3 sound to cause severe consequence on PCC enzyme, but apparently Patient 3 has better phenotype (especially related with her cardiac condition). Do the authors have any explanation/opinion on this?

Minor corrections:
- Please revise all typos, bad sentences/grammar, the wrong use of capital and italic, and space on some parts
- Please be consistent on how to write the references.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

No

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Acceptable

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